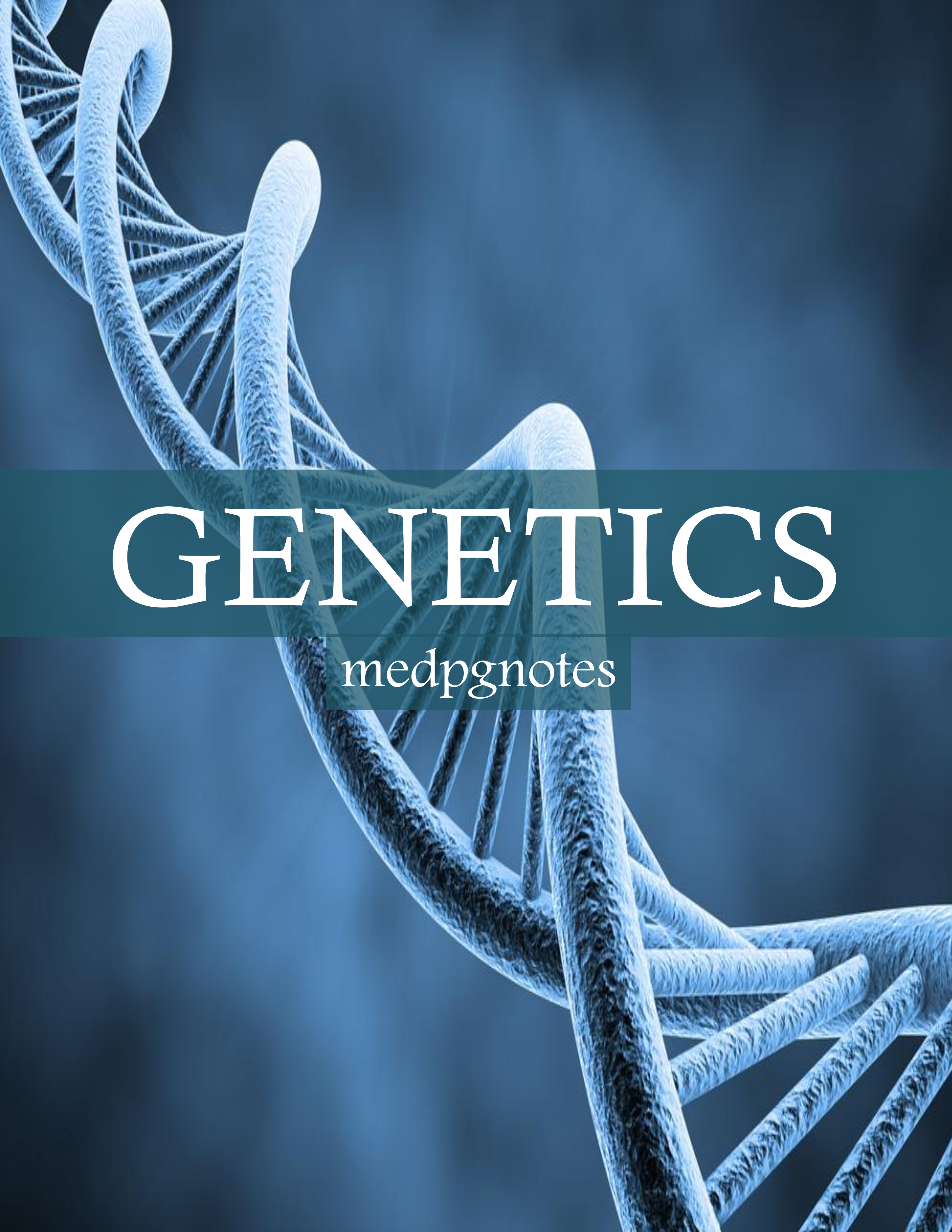

GENETICS

medpgnotes

GENETICS

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KEY TO THIS DOCUMENT

Text in normal font – Must read point. Asked in any previous medical entrance examinations

Text in bold font – Point from Harrison's text book of internal medicine 18th edition

Text in italic font – Can be read if you are thorough with above two.

GENERAL FEATURES OF GENES AND CHROMOSOMES

GENERAL FEATURES OF GENETICS

Human DNA	**3 million base pairs**
Polymorphic microsatellite marker also known as	**Short tandem repeats**
Human genome project is completed in	**May 2006**
Hap map project for	**Single nucleotide polymorphism**
Almost all single nucleotide polymorphism have	*2 alleles (biallelic)*
Gene rearrangement classically occurs in	*Immunoglobulin gene*

GENERAL FEATURES OF CHROMOSOMES

Components of chromosome	DNA, histone
Long and short arms of chromosomes	Q and p respectively
During mitosis, anaphase is associated with	*Separation of sister chromatids to form chromosomes*
Y chromosome	Acrocentric
SRY gene located on	Short arm of Y chromosome
Gene coding androgen receptors are located in	Long arm on X chromosome
Acriflavin Schiff	*X chromosome (Feulgen reaction)*
Quinacrine staining	*Y chromosome*
Increased in Prader Willi syndrome	Ghrelin
Maternal disomy of chromosome 15 is seen in	Prader willi syndrome
MC mechanism of Prader Willi syndrome	*Trisomy rescue*
Trisomy rescue is seen in	*Chromosome 15*
Prader Willi syndrome is associated with	Hypotonia, obesity
Features of Prader Willi syndrome	*Short stature, narrow bifrontal diameter*
MC mechanism of Angelmann syndrome	*Monosomy rescue*
If a chromosome divides in an axis perpendicular to its usual axis of division it is going to form	Isochromosome
Transfer of genetic material in 1 meiotic division between 2 non homologous chromosome	Translocation
Crossing over	*Between non sister chromatid of homologous chromosome*
Cross over	*Pachytene stage of prophase I of meiosis*
Highly condensed chromosome in	*Metaphase*
Pericentric inversion	*Involves 2 arms*
Paracentric inversion	*Involves 1 arm only*
Persons with inversion, insertion and translocation	*Phenotypically normal, increased risk of miscarriage*
Deletion is associated with	*Mental retardation*
Telomere mutation is associated with	*Dyskeratosis congenital, aplastic anemia, pulmonary and hepatic fibrosis*

Chromosomal instability syndromes	*Nijmegen syndrome, ICF syndrome (Immunodeficiency, centromere instability, facial anomalies), Robert's syndrome*
Additional X chromosome is associated with	*Low IQ*
Additional Y chromosome is associated with	*Aggressive behavior*

BARR BODY

Barr body is first detected in	*Brain*
Barr body	*Facultative heterochromatism*
Barr body	*Buccar smear, saliva, hair follicle*
Barr bodies are demonstrated in	Neutrophils
Barr body is found in	Interphase
Barr bodies are	Squamous epithelial cell
Barr bodies are NOT seen in	Turner syndrome
Sex can be established by examining hair root cells for	Barr body

GENES

Gene is	*Cistron*
Genes	Smallest functional unit of genome, Promoter and enhancer genes are typical example, Cistron is single functional unit
Gene	Repressor binds operator gene, Regulator genes produce repressor subunit, IP TG is inducer but not substrate, Regulator gene is NOT inducible
Gene whose phenotype is expressed	*Epistatic*
NOT true about eukaryotic gene	Polycistronic mRNA, Non coding Exon
Differential expression of same gene depending on parent of origin	Genomic imprinting
Functional gene is inherited from one parent	Genomic imprinting
Gene regulating normal morphogenesis during development	Homeobox gene
Approximate number of genes in human genome	20,000 – 25,000
ALU family	*5-6% of genome*
Coding region	*Less than 1.1% of genome*
Highly repetitive means	*More than 10^6 times*
Process underlying differences in expression of a gene according to which parent has transmitted	Anticipation
Small head, minor anomalies of face, growth delay and developmental delay, NOT have	Polygenic syndrome
Punnet square is used to	Predict genotype of offspring
Methods of fusing two cells in genetic combination	Ethylene glycol, electric current, viral transformation
A person showing two cell lines derived from two	Chimerism

different zygotes	
Atavism	Child resembling grandparents
DNA methylation	Alters gene expression, Genetic code remains intact, Role in carcinogenesis, Protective mechanism against cleavage by restriction endonuclease
Single gene defect causing multiple unrelated problems	Pleotropism
Inactivation of X chromosome	*Lyonization*

ANEUPLOIDY

Primary defect in a structure	Malformation
Aneuploidy due to	Non disjunction at meiosis
NOT true about aneuploidy	Occurrence of aneuploidy has no relation with progression of mother age
NOT true about aneuploidy	50-60% of trisomy 18 fetus dies between 12 and 40 weeks
NOT used for detection of specific aneuploidy	Microarray
Most sensitive sonological indicator for aneuploidy	Nuchal transluscency

CHROMOSOMES

Chromosome 1	Steroid resistant nephrotic syndrome (NHS2 – podocin)
Chromosome 3	Clear cell renal carcinoma, VHL gene
Chromosome 4	*Huntington's chorea*
Chromosome 5	APC gene
Chromosome 6	Major histocompatibility complex, HLA 6p
Chromosome 7	Cystic fibrosis
Chromosome 9	*Tuberous sclerosis*
Chromosome 11	*Ataxia telangiectasia gene*
Chromosome 12	*Darrier disease*
Chromosome 13	Hereditary retinoblastoma - deletion of chromosome 13q14, Wilson's disease, BRCA 2, Friedrich's ataxia
Chromosome 17	BRCA 1, P53
Chromosome 18	Edward's Syndrome
Chromosome 19	**Myotonic dystrophy, JAK, NPHS1 finnish type (nephrin), Peutz Jegher syndrome**
Chromosome 20	*Prion disease*
Chromosome 21	Down syndrome, Gene for folate carrier protein

CYTOGENETICS AND DNA RECOMBINANT TECHNOLOGY

GENERAL FEATURES OF CYTOGENETIC STUDIES

Test using oligomer with single base pair substitution	RFLP
Linkage analysis	Detection of characteristic DNA polymorphism in a

	family is associated with disorders
Phage DNA	Restriction enzyme sites
DNA reconstruction technology	Restriction endonucleases are involved, DNA ligase is used, Reverse transcriptase is needed
Enzymes used in DNA research programme	Polymerase, Exonuclease, Nuclease, Alkaline phosphatase
Disputed maternity is solved by	Blood grouping, HLA, DNA fingerprinting
NOT used for gene sequencing	*Satellite DNA*
Chromosomal mutation cannot be detected by	*Agarose gel electrophoresis*
Methods to induce cell fusion	*By adding some inactivated virus to surface membrane, by adding polyethylene glycol, by mild electric shock*

POLYMERASE CHAIN REACTION

DNA amplification is done in	PCR, NASBA, Ligase chain reaction
Advantage of NASBA over PCR	Does not require thermal cycling
Polymerase chain reaction	Kary Mullis
Polymerase chain reaction	Carried out by thermostable DNA polymerase, Exponential, Specific, Primer is needed, Enzymatic DNA amplification, Annealing is done after DNA denaturation, Detection of infecting organism, DNA polymerase
RT PCR used for	DNA Amplification
PCR is used to amplify	Small segment of DNA
Enzyme used in PCR	DNA polymerase
Cation used in PCR	Magnesium
Polymerase used in recombinant synthesis	Thermostable, Replicates dsDNA
In PCR, aquaticus thermophilus is preferred over E.coli because	Thermostable at temperature at which DNA liquefies
Primer in PCR are complementary to	*3' end of both the strands*
Used in PCR	Mg++
NOT required for PCR	Radiolabelled DNA probe, Dideoxynucleotides
NOT true about PCR	Required atleast 1 week time for synthesis, DNA polymerase has to added on each cycle
NOT a requirement of PCR	Restriction endonuclease
SYBR green dye is used for	PCR
Effective polymerase chain reaction was repeated for 3 cycles on a DNA molecule. resulting formation of copies	8 times

RESTRICTION FRAGMENT LENGTH POLYMORPHISM

Function of endonuclease	Cut DNA at specific DNA sequence
Restriction endonuclease	Cut both strands of dsDNA, Cut ends produced are sticky, Cut ends produced are blunt, Recognize short sequence of DNA
What will occur if bacteria acquires genes coding for	Bacteria will die as it lacks methylase

restriction endonuclease	
Restriction endonucleases are derived from	Bacteria
Enzyme required to maintain DNA library by breaking a DNA into piece	Restriction endonuclease
Restriction endonucleases are used in	RFLP
Restriction enzymes	Palindromic, Produces DNA sticky end, Restrict replication of DNA, Breaks at sugar phosphate bond, Protects bacteria from infection by virus, Restrict replication of DNA
Most characteristic function of type II restriction enzyme	Cut DNA at palindromic site
After digestion by restriction endonuclease DNA strands can be joined by	DNA ligase
Bacteria acquire restriction endonuclease by horizontal transfer. the result would be	Bacteria cannot survive because it lacks DNA methylase that protects host DNA
Blunt end producing restriction endonuclease	*Hpal*
Restriction fragment gene	Detected by southern blot, Used for identification of gene for genomic mapping, RFLP is a DNA variation sequence
DNA fragments formed by action of restriction endonucleases are separated by	Agarose gel electrophoresis
DNA restriction is done by	Electrophoresis agar gel method
Polymorphism	Single phenotype, single locus = multiple normal alleles
Restriction fragment length polymorphism is used for	Analysis of chromosome structures
RFLP	Detects mutation, Recognizes trinucleotide repeats, Detects deletion, Blunt ends are produced

DNA FINGERPRINTING

DNA fingerprinting was founded by	Jeffrey
DNA fingerprinting is based on possessing in DNA of	Variable number tandem repeats
DNA fingerprinting is done by	Splitting of DNA, DNA of WBC, DNA from nucleated cells
Sample for DNA testing	Femur and bone marrow without preservative
In DNA testing, tissue sample taken in an autopsy	Spleen
Blood samples for DNA fingerprinting is stored in	EDTA
Microsatellite sequence	Short sequence (2-5) repeat DNA

FLUORESCENCE AND LUMINENCE

Fluorescence means	Release of longer wave length light on absorbing light of shorter wavelength
Chemiluminence	Excited electron in higher orbit comes to lower orbit by emitting energy in the form of photon Bioluminence form of chemiluminence
Photoluminence	Absorbed radiation emitted as light
Electrophotochemiluminence	Light emitting reaction which take place by use of electrical current

Fireflies produce light due to	ATP
Nephelometry is based on the principle of	Light attenuated in intensity by scattering
DNA estimation can be done by	Spectrophotometer

KARYOTYPING

MC Karyotyping technique	G band (Giemsia)
Procedure for karyotyping under light microscopy	G banding
Karyotyping, Chromosomes arrested in	Metaphase
Karyotyping technique for terminal ends	R banding
Karyotyping technique for Centromeric region	C banding
T banding is used for	*Telomere*
Samples taken in a macerated baby for detecting abnormality in Karyotype	Fibroblast from Skin fascia
Karyotyping of fetus can be done from	Lymphocyte, amniocyte, fibroblast
Karyotyping of fetus cannot be done from	Fetal skin biopsy
Karyotyping is useful in diagnosis of	Chromosomal abnormalities
Karyotype is	Size, shape and number of chromosome
Banding is a	Method of detecting single gene deletion
Absent thumb, radial deviation of wrist, bowing of forearm. Needed investigation	Karyotyping

BLOTTING

DNA detected by	Southern blot
Highly sensitive method for identifying DNA fragments by DNA-DNA hybridization	Southern blotting
Western blot	Separation of proteins are bases on mass, SDS PAGE electrophoresis is used for separation, Enzyme linked antibodies are used, Confirmatory in AIDS
Probe used in western blot	Antibody
Antigen antibody reaction is detected by	ELISA, Western blot
When compared to western blot, ELISA is	More sensitive, less specific
Western blotting technique is done for	Protein
Northern blot is used for separation of	mRNA

HYBRIDOMA TECHNOLOGY

Monoclonal antibody	Produced by hybridoma technology, Requires in small quantity, Used for blood grouping
Hybridoma refers to	Technique for raising monoclonal antibodies
Hybridoma	Immortalize myeloma cell, Prior immunization is done, Produce monospecific and monoclonal antibody
Hybridoma technology	Specific antibody producing cells are integrated with myeloma cells. Aminopterin, a folate antagonist inhibits de novo pathway, HGPRTase and thymidylate synthase are required for salvage pathway

Hybridoma technique is used to approve	*Purified antibodies*

GENETIC STUDIES

Difference in gene expression is studies by	Microarray
Technique used for detection of variation in DNA sequence and gene expression	Microarray
Methods used for detection of gene expression	RT PCR, cDNA microarray, northern blot
Investigation of choice for identifying genetic loci	FISH
For measurement of long DNA molecules (50-100 kb) following method is used	RFLP
Gene library	DNA nucleotide of fragment, Complete collection of gene copies of one organism
Subtelomeric rearrangement of genes identified by	FISH, MAPH, cGH assay
Mutation that completely disrupt the function of gene used in	Knock out, Targeted gene disruption

CHROMOSOMAL STUDIES

Rapid method of chromosome identification in intersex	FISH
Chromosomes are visualized through light microscope with resolution of	10Mbp
Nanotechnology is used in Chromosomal study because	Narrow emission spectra
Test to differentiate in the chromosome of normal & cancer cells	Comparative genomic hybridization

TRANSGENIC ANIMALS

Transgenic mice	Developed from DNA insertion into fertilized egg, Have same genome as parents except one or more genes, Homozygous are selected
Most suited method to assess function of gene	Transgenic animals
Nude mouse is used in genetics because of	Absence of thymus, Xenograft can be sustained for weeks

GENE INCORPORATION

Methods for producing fusion of cells	Attaching inactive viral particles on cell membrane, Adding polyethylene glycol, Applying small electric current
In gene cloning, large fragment can be incorporated in	Cosmid
In DNA transfer the vectors used from smallest to largest is	Plasmids, bacteriophage, cosmid, bacterial artificial chromosome, yeast artificial chromosome, human artificial chromosome
Vector for cloning of gene more than 500 kb	*YAC*

Cloning vector has least insert size	*Plasmid pbr*
Most common marker used in gene cloning	*Beta galactosidase gene*
Microorganism used as vector for transforming plants	*Agorabacterium tumifaciens*
NOT a method of introducing gene in target cells	FISH
Movement of protein from nucleus to cytoplasm can be seen by	FRAP
In molecular cloning blue white screening is used for	To identify desired chromosomal DNA insert in plasmid vectors

RECOMBINANT TECHNOLOGY

Site specific recombination	RE+ ligase
Strategy used to increase the yield of protein produced in recombinant protein synthesis	Genes for protease inhibitors
Process in a vector is used to yield of protein produced in recombinant protein synthesis	Promoter induction
Starting material for production of insulin from bacteria	mRNA from beta pancreatic cell of human

GENE THERAPY

Purpose of gene therapy	Replacement of abnormal gene by normal gene
Gene therapy methods are	Electroporation, Intranuclear injection, Site directed mutagenesis, Retrovirus
First gene therapy (somatic enzyme) was successfully done in	SCID (ADA deficiency)
Gene therapy given for	Cystic fibrosis
Evaluate a function of a gene by introducing a genetic material is	Gene therapy

MODE OF INHERITANCE OF DISEASES

INHERITANCE OF DISEASES

AUTOSOMAL DOMINANT	AUTOSOMAL RECESSIVE
Hereditary spherocytosis	Cystic fibrosis
Achondroplasia	Wilson disease
Huntington's chorea	Fanconi syndrome
Familial hypercholesterolemia	Phenylketonuria
Retinoblastoma	Alpha 1 antitrypsin deficiency
Von willebrand disease	Homocystinuria
Myotonic dystrophy	Ataxia telangiectasia
Otospongiosis	Thalassemia

Marfan syndrome Familial adenomatous polyposis Von Hippel Lindeau syndrome Neurofibromatosis	Sickle cell disease Juene disease

X LINKED DOMINANT	X LINKED RECESSIVE
Incontinential pigmenti Alport Syndrome Rett Syndrome Vitamin D Resistant rickets	G6PD deficiency Duchene muscular dystrophy Fragile X syndrome Kinky hair disease (Menke hair disease) Fabry disease

FEATURES OF INHERITANCE

Loss of heterozygosity means	Loss of normal allele in mutant gene
Father has some abnormalities, one of the three children has similar features. Mother is normal. least likely pattern	Autosomal dominant
MC group of diseases following mendelian inheritance	Autosomal dominant
Male to male transmission is a key feature in	*Autosomal dominant*
An affected male infant born to normal parents could NOT be an example of	Autosomal dominant disorder
True regarding autosomal dominant inheritance	1:2,1:3,1:1
Autosomal dominant disease	*Incomplete penetrance, variable expression*
Autosomal dominant inheritance	**Incomplete penetrance**
Dominant negative inheritance	Marfan syndrome, Ehler Danlos syndrome, Osteogenesis imperfecta
A parent is homozygous and a parent heterozygous for an autosomal recessive gene. outcome	50% affected rest are carriers
For a normal husband and wife, the first child was diagnosed to have cystic fibrosis. percentage of chances for the second child be affected	0
An albino girl gets married to a normal boy. what are the chances of their having an affected child and what are the chances of their children being carrier	None affected, all are carrier
Parents are clinically normal and only siblings are affected but males and females are affected in equal proportion	Autosomal recessive
Expression in Autosomal recessive inheritance	Uniform
A particular genetic disorder appears in three consecutive generations of a family without any sex predilection. It was also noticed that phenotypically normal family members were having healthy offspring. pattern of inheritance	Uniparental disomy
Father to son inheritance is never seen in case of	X linked recessive inheritance
Males are commonly affected than females in	X linked recessive inheritance
X linked recessive disease	50% male affected if female is carrier male unaffected
NOT true about X linked recessive disease	Male to male transmission only

NOT an X linked recessive	Rett syndrome
NOT true about x linked recessive	Unaffected males do NOT transmit trait to any offspring
Male to male transmission in	*Y linked inheritance (Leri Weil dyschondrosteosis, Langer mesomelic dwarfism)*
Non mendelian inheritance	Genomic imprinting, Uniparental disomy, Mitochondrial inheritance
Couple has two children affected with tuberous sclerosis, both parents normal. which explains two affected children in family	Gemline mosaicism
Two siblings with osteogenesis imperfecta have normal parents. mode of inheritance is explained by	Germline mosaicism
Difference in expression of gene in sibling as inherited by father termed as	Mosaicism
Presence of two or more genetic cell lines in tissues of individual	**Mosaicism**
Germline mosaicism is seen in	Tuberous sclerosis, Osteogenesis imperfecta
Ability of the stem cells to cross barrier of differentiation to transform into a cell of another lineage expressing the molecular characteristics of different cell type with the ability to perform function of new cell type	Transdifferentiation

MITOCHONDRIAL INHERITANCE

Mitochondrial DNA is known for	Maternal inheritance, Heteroplasmy, Leber hereditary optic neuropathy is prototype
NARP syndrome is seen in	Mitochondrial diseases
Leigh's disease, Leber's hereditary optic atrophy, Myoclonic epilepsy	Mitochondrial inheritance
Pearson syndrome	**Pancreatic insufficiency, pancytopenia, lactic acidosis**
Pearson syndrome is associated with	*Sideroblastic anemia*
NOT a mitochondrial disease	Nemaline myopathy

GENETIC DISORDERS

GENERAL FEATURES OF GENETIC DISORDERS

Abnormality in total chromosome number	Mongolism, gonadal dysgenesis, Klinefelter syndrome
In Robertsonian translocation fusion occurs in	*Centromeres*
Tetrasomy 12	*Pallister Killian syndrome*
Uniparental disomy is NOT seen in	Bloom syndrome
FGFR2 mutation results in	**Crouzon syndrome (Craniofacial synostosis), Pfeiffer syndrome (Acrocephalopolysyndactyly)**
Trinucleotide repeat disorders	**Fragile X syndrome (CGG, GCC), Myotonic dystrophy (CTG),**

	Friedrich Ataxia (GAA), Other disorders (CAG)
MC trisomy	**Trisomy 16**
MC cause of trisomy	**Non disjunction at meiosis I**
Intrachromosomal deletion syndrome	**Wolf Hirschorn syndrome, Cri du chat syndrome**
Microdeletion	**Miller Dieker syndrome, Smith Magenis syndrome**
4p deletion is associated with	*Wolf Hirschhorm syndrome (Greek helmet fever)*
Menky Kinky hair disease is due to	*Defective copper transport*

REARRANGEMENT OF TRANSCRIPTION FACTORS

TRANSCRIPTION FACTOR CLASS	EXAMPLE	ASSOCIATED DISORDER
Homeobox	**IPF1**	**MODY 4**
Forkhead	**HNF 4 alpha, HNF 1 alpha, HNF 1 beta**	**MODY 1,3,5**
Leucine zipper	**Retinal leucine zipper**	**Autosomal dominant Retinitis pigmentosa**
Basic helix loop helix	**MITF**	**Waardenberg syndrome type 2A**
Zinc finger protein	**WT1**	**WAGR**

DOWN'S SYNDROME

A 35 years old lady has chromosomal translocation 21/21. Risk of down's syndrome in the child is	100%
Down's syndrome most commonly occurs due to	Non disjunction in maternal meiosis
Down's syndrome in advanced maternal age is most commonly due to	Non disjunction
MC genetic cause of repetitive Down's syndrome in young mother	*Robertsonian translocation*
Only genetic defect with 100% transmission in Down's syndrome	*Isochromosome 21 (21q, 21q)*
Chromosomal anomaly in mongolism	Trisomy 21
Disease associated with increased age of father	Mental illness
MC trisomy	Trisomy 21
Increased nuchal fold thickness	Down syndrome
Down's syndrome associated with	Duodenal atresia, Hirschprung disease
Shape of head in Down's syndrome	Brachiocephalic
Third fontanelle is seen in	*Down's syndrome*
Epicanthal fold, broad nose, protruding tongue	*Down's syndrome*
Speckled iris	*Brushfield spot*
Anterior scalloping of vertebral body	*Down's syndrome*
Memory impairment most likely in	Down syndrome
Differentiation between Down's syndrome and Cretinism	Wide set eyes
Down's syndrome predispose to	AML
Hall's criteria for	*Down's syndrome*
NOT a feature of Down's syndrome	Hypertonicity
NOT a characteristic of Down's syndrome	Prominent occiput

NOT a feature of Down's syndrome	Undescended testis
G2 female 9 weeks pregnant, had a previous child with down's syndrome. She doesn't want another child with Down's syndrome. you will tell her	Chorionic villous biopsy at this stage will confirm presence or absence
Screening for down syndrome in which age group of pregnancy	35 years
Best serological marker for diagnosis of down's syndrome in 1st trimester of pregnancy	Beta hcg
Triple test	Beta hcg, AFP, Serum estradiol
Quadruple test for Down's syndrome	AFP, unconjugated estradiol, HCG, Inhibin A
PAPPA in down's syndrome	Decreased
NOT included in triple screening of Down's syndrome	Acetylcholinesterase
NOT a biochemical marker in triple test	Human placental lactogen
NOT a diagnostic marker for Down's syndrome	Progesterone
In down's syndrome patient posted for surgery, necessary pre operative investigation	Echocardiography
Nuchal transluscency at 14 weeks	Down's syndrome
Ultrasound marker associated with greatest risk for Trisomy 21	Nuchal edema
Measurement of nuchal translucency is best made between	11 and 14 weeks of gestation
Advantages of ultrasound nuchal transluscency over biochemical screening for Down's syndrome	*Better in multiple gestation*
Features of Down's syndrome associated with first trimester	*Absent nasal bone, increased nuchal translucency, PAPP is decreased*
Investigation done in first trimester for diagnosis of Down's syndrome	*Chorionic villi sampling*
Best way to identify Down's syndrome in second trimester	*Amniocentesis*
Single umbilical artery NOT associated with	Advanced maternal age
NOT a USG marker of Trisomy 21	Single umbilical artery
NOT a feature of second trimester USG of Down's syndrome	Choroid plexus cyst
NOT a feature of down's syndrome	Increased PAPPA
Increased Maternal age is associated with	Down's syndrome, XO, XXY

TRISOMY 13

Trisomy 13	Patau syndrome
Child presenting with multiple deformities, cleft lip and cleft palate, microcephaly, small eyes, scalp defect and polydactyly	Trisomy 13
Multiple deformities, cleft lip, cleft palate, microcephaly. small eyes, scalp defect, polydactyly	Trisomy 13
MC ocular manifestation in Trisomy 13	Bilateral microphthalmos
Patau syndrome is associated with	*VSD*

TRISOMY 18

Edward syndrome	Trisomy 18 (Rocker bottom foot)
Edward syndrome	Low set ears, Rocker bottom foot, Renal malformations
Index finger overlapping 3rd finger, little finger overlapping 4th finger	*Edward syndrome*
Features of Edward syndrome	*Mental retardation, stunting of growth, congenital disease*
NOT a feature of Edward syndrome	*Hypotonia*

TRISOMY 22

Cat eye syndrome	Partial trisomy 22

TURNER SYNDROME

Turner syndrome	*Hypergonadotrophic hypogonadism*
Turner syndrome	*Lost X chromosome of paternal origin, mosaicism of X chromosome, prophylactic gonadectomy*
Turner syndrome	Edema of hands and foot, Streak ovaries, XO, Infertility , Coarctation of aorta
XO is characterized by	Short stature, Shield chest, Cubitus valgus, Edema over hand and feet
XO chromosomal defect	Short stature, Infertility, Webbed neck, Widely spaced nipples
Webbing of neck, increased carrying angle, short fourth metacarpal, posterior hairline	Turner syndrome
20 years female, primary amenorrhoea, absent breast, hypoplastic uterus	Turner's syndrome
19 year female, short stature, wide spread nipple and primary amenorrhoea	45X
Turner syndrome	Normal intelligence
Cystic hygroma in USG is suggestive of	Turner syndrome
Webbing of neck, increased carrying angle, low posterior hair line, short fourth metacarpal	Turner syndrome
Turner's syndrome is maximally associated with	Coarctation of aorta
Turner's syndrome represented by	XO
Karyotyping in Turner syndrome	45X
NOT true about Turner's syndrome	Occurrence influenced by maternal age
NOT true about Turner's syndrome	Tall stature

KLINEFELTER SYNDROME

Klinefelter syndrome	XXY, Male hypogonadism, FSH increased

47 XXY with hypogonadism	Klinefelter syndrome
Disorder of sex chromosome	Klinefelter syndrome
Features of Klinefelter's syndrome	Gynecomastia, sterility, small testis with atrophy, azoospermia
Sexually underdeveloped male, rudimentary testis, prostate, sparse facial and pubic hair, long arms legs, large hands and feet	46 XXY
Hypergonadotrophic hypogonadism	Klinefelter syndrome, Viral orchitis, Noonan syndrome
MC cause of hypergonadotrophic hypogonadism in males	Klinefelter syndrome
Klinefelter syndrome	*They are 20 times as likely as normal men to develop breast cancer*
Testis in Klinefelter syndrome	*Small firm testis*
Young male with delayed puberty with decreased FSH, LH and testosterone (hypogonadotrophic). NOT possible	Klinefelter syndrome
Death in Klinefelter syndrome is due to	Infection

NOONAN SYNDROME

Noonan syndrome	Affects male and female, Short stature, ASD
Antimongoloid slant, pulmonary stenosis, short stature, undescented testes	Noonan syndrome
MC cardiac abnormality in Noonan syndrome	Pulmonary stenosis
NOT true about Noonan syndrome	Infertility in females, Chromosomal abnormality

DISORDERS OF DNA REPAIR MECHANISM

Diseases having defect in DNA repair mechanism	Xeroderma pigmentosa, Fanconi syndrome, Hereditary non polyposis colon cancer
Congenital syndromes having lymphoproliferative malignancy	Bloom syndrome, Fanconi anemia, Chediak higashi syndrome, Ataxia telangiectasia, Klinefelter syndrome, Kostman syndrome
Cutaneous manifestation occurring in all DNA repair defect	*Café au Lait macules*
Bloom syndrome	**Helicase deficiency**
Features of bloom syndrome	*Photosensitivity, bullous eruption on lips, café au lait spot*
NOT true about bloom syndrome	*Common in elderly*
NOT true about Bloom syndrome	IgA absent
Chromosomal breakage syndrome	Fanconi anemia, Bloom syndrome, Ataxia telangiectasia
Ataxia telangiectasia	Chronic sinopulmonary disease, chromosomal breakage, IgA deficiency, IgG2 deficiency
Gene instability associated with malignancy in	Ataxia telangiectasia
Ataxia telangiectasia	Ovarian dysgenesis, Recurrent Sinopulmonary Infections, Cerebellar Ataxia
Defective protein in ataxia telangiectasia	*DNA dependent protein kinase*

Ataxia telangiectasia mutated (ATM)	*DNA dependent protein kinase activates ku helicase*
Cause of death in ataxia telangiectasia	*Malignancy*
Mismatch repair	*HNPCC*
Endonuclease of mismatch repair is known as	*GATC endonuclease*
Nucleotide excision repair	*Xeroderma pigmentosa*
Endonuclease of base excision	*Apurinic or apyrimidinic endonuclease*
Nucleotide excision	*Excinuclease*
Defect in xeroderma pigmentosa	*DNA ligase*
dsDNA break repair	*Ataxia telangiectasia, Bloom syndrome, Fanconi syndrome*